Add-On Event Descriptions

Please see more information in ASHG's Online Planner.

Receptions & Luncheons

Trainee Reception (open to trainee and early career attendees only)

Wednesday, November 1: 7:30 pm - 9:30 pm

Pricing: \$35 ASHG member, \$50 nonmember. Advance ticket purchase required to attend.

Includes heavy appetizers and one drink ticket (beer, wine, and soft drinks only).

This event will give you the opportunity to connect with other meeting attendees in person while having a good time! This relaxed, informal, and interactive networking event will help you meet existing and potential collaborators, make new friends, and foster new connections. There will also be a chance to win prizes, like 1 year of ASHG membership, by playing games. With your event ticket you will receive appetizers as well as one drink ticket.

Diversity, Equity, and Inclusion Luncheon

Thursday, November 2: 12:15 pm - 1:45 pm

Pricing: \$35 ASHG member, \$50 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: Gary H Hibbons; TBD.

The Diversity, Equity, and Inclusion Luncheon will feature as a keynote speaker, Gary H. Gibbons, M.D., Director of the National Heart, Lung, and Blood Institute at the National Institutes of Health. Dr. Gibbons will highlight how leveraging inclusive excellence in genomics research can advance scientific discovery and innovation across heart, lung, blood, and sleep diseases or disorders and improve health outcomes for all. During the event, panelists Athena Davenport, Ph.D. and Paola Giusti Rodriguez, Ph.D., both inaugural Human Genetics Scholars, will share their perspectives, expertise, and experiences as early to mid-career researchers in the field. The luncheon provides an exceptional venue for networking, and the audience is encouraged to bring questions and lend your input to this important discussion.

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The Diversity, Equity, and Inclusion Reception scheduled for Thursday, November 2, will recognize and celebrate the importance of diversity, equity, and inclusion, as well as provide networking for the research and training community.

Race, Ancestry, Both, or Neither?: Introducing Frameworks for Population Descriptors in Genomics Research

Friday, November 3: 12:15 pm – 1:45 pm

Pricing: \$35 ASHG member, \$50 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: V. Reid Sutton; Genevieve Wojcik; Vence Bonham, Jr.; & Shoumita Dasgupta.

This session introduces junior investigators and trainees to emerging questions and strategies for considering whether, why, and when to use population descriptors (e.g., race, ethnicity, and ancestry) in genomics research. Recent conversations in the field have raised awareness about the uses and misuses of descriptors and highlighted key areas for potential actions across organizations and sectors. How can researchers begin to rethink their own assumptions and integrate new frameworks into different types of research studies? What can departments do to help trainees begin to explore these questions? Join researchers and educators in this roll-up-our-sleeves session to consider and rethink how and why we use population descriptors in genomics research.

The Confidence Factor: Thriving in Your Career

Friday, November 3: 12:15 pm - 1:45 pm

Pricing: \$35 ASHG member, \$50 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: TBD.

This panel event is designed for early career professionals seeking to gain confidence and unlock their career potential. During this 90-minute session, you'll hear from 3 experienced professionals in genetics who will share their personal journeys to gaining confidence, as well as practical tips for achieving your career goals. The event will feature both panel presentations and round table speed discussions, allowing you to connect with your peers and gain insights into the connection between confidence and success.

Behind-the-Scenes: Publications Workshop

Saturday, November 4: 12:00 pm - 1:00 pm

Pricing: \$35 ASHG member, \$50 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: AJHG and HGG Advances editors.

This workshop being held on Saturday, November 4, will provide you with information to help navigate the scientific publication process. Editors from multiple journals, including AJHG and HGG Advances, will give you a behind-the-scenes view of scientific publishing, covering topics such as data sharing, open access publication, and the ins and outs of the peer review process. There will be plenty of time for questions and networking. This session is geared towards trainees and junior faculty who are relatively new to publishing.

ASHG's 75th Anniversary Gala Celebration

Saturday, November 4: 7:00 pm - 10:00 pm

Pricing: \$85 ASHG regular member; \$40 trainee member; \$95 nonmember. **Advance ticket purchase required to attend.**

Registered attendees may now purchase up to 4 tickets! Note: Due to the nature of the facility, we can't accommodate children under the age of 16.

Includes heavy appetizers and two drink tickets (beer, wine, and soft drinks only).

Join us for a special closing gala reception at the ASHG 2023 Annual Meeting honoring the Society's 75th Anniversary on Saturday, November 4, 7:00 pm - 10:00 pm at the Ronald Reagan Building and International Trade Center. Themed "One Humanity, Many Genomes," the event will spotlight 75 years of innovation in the field and how genetics and genomics research is generating benefits for people everywhere. The evening will include drinks, food, and live music from Ethidium Spill featuring ASHG members Francis Collins, Anthony Antonellis, and Elliott Marguilies. Join us in celebrating human genetics, our research community, and Society's bright future leading new discoveries and better health for people everywhere. See additional info here.

Workshops

Teaching Variant Curation Through Team-based, Active Learning Approaches

Wednesday, November 1: 11:00 am – 1:00 pm

Pricing: \$35 ASHG member, \$50 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: Barbara Fortini & Emily Quinn.

Single nucleotide variant (SNV) curation is an important skill to develop for students in genetic counseling, genomics, and bioinformatics master's programs. It is also a common subject for continuing education workshops for professionals working in clinical genetics roles. While the number of evidence categories and scoring criteria can be intimidating for new learners, the process lends itself well to team-based, active learning approaches in the classroom. This workshop will use the ACMG/AMP standards and guidelines for germline SNV interpretation as a case study for developing classroom activities that engage learners, develop variant analysis skills, and build confidence in the curation process for students.

Throughout the workshop, participants will gain hands-on experience designing instructional activities for clinical genetics concepts, assessing the effectiveness of the activities, and constructing an instructional scaffold to guide learners to mastery of the curation process. Participants will also discuss the benefits of teaching in a team-based, active modality. While the workshop will focus on in-person educational formats, there will also be discussion of how to adapt the same activities to synchronous, online delivery and self-paced, solo learners.

Learning Objectives

- Design team-based, active learning activities for teaching genetics concepts.
- Assess the effectiveness of active learning activities.
- Plan an educational approach to develop students' skills in SNV interpretation.
- Describe the benefits of team-based learning approaches for students.

Genomic Analysis in the All of Us Researcher Workbench

Wednesday, November 1: 11:00 am - 1:00 pm

Pricing: \$35 ASHG member, \$50 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: Alexander Bick & Robert Carroll.

The NIH's All of Us Research Program is committed to the ambitious mission of collecting multiple types of health data from a million or more participants to create a diverse research resource that accelerates precision medicine. The All of Us Researcher Workbench is a secure, cloud-based platform where registered researchers access and analyze data from over 413,000 program participants. Of these participants we will have released >245,000 whole genome sequenced samples and >314,000 genotyping arrays in the All of Us Researcher Workbench by Spring of 2023. By Fall of 2023, we also anticipate the availability of the first set of long read genome sequencing data. This genomic data is combined with many types of phenotypic and auxiliary data types including electronic health records, survey data, physical measurements, mobile health data (Fitbit). This free-to-access platform is also a space where researchers have the ability to work collaboratively with other registered researchers through shared workspaces. In addition to increasing the number of samples available to researchers since its launch of genomic data in March 2022, the All of Us program has expanded the resources available for researchers to explore the genomic data. All of Us has both added new analysis tools, such as REGENIE, and expanded the data types it provides by introducing structural variant datasets from 10,000 participants and 1,000 long read samples in Spring 2023. A main goal of the program is to make this data accessible to a broad array of researchers. The evolution of genomic research is transforming the treatment of disease, motivating wellness and health promotion, and modernizing disease prevention in public health.

This workshop invites participation from researchers interested in learning how to utilize the All of Us dataset and Researcher Workbench for their own research. This session will present an overview of the Researcher Workbench, highlighting the expanded data with the Spring 2023 release and then 1) provide an interactive, guided demonstration of the Researcher Workbench, 2) provide participants a hands-on opportunity to replicate a research study with genomics data using newly added tools within the Workbench, 3) conduct an interactive Q&A session to engage attendees about All of Us Researcher Workbench and the novel technology implemented to conduct genomic analyses, and 4) discuss the approach to scale genomics analysis to 1 million participants.

Learning Objectives

- Identify the key cloud computing concepts that enable All of Us Researcher Workbench analysis.
- Construct a phenotype from All of Us participant data in the All of Us Researcher Workbench.
- Design a genetic association study leveraging All of Us data in the All of Us Researcher Workbench.
- Describe how short and long read sequencing data differ from each other.

Using UK Biobank to Scale up your Research

Wednesday, November 1: 2:00 pm - 4:00 pm

Pricing: \$35 ASHG member, \$50 nonmember. **Advance ticket purchase required to attend.** Includes light snacks and drinks.

Speakers: Aleksejs Sazonovs; Jeffrey Barrerr; Laura Fachal; & Sini Kerminen.

Biobank scale datasets have become an important part of nearly all aspects of health research. By systematically linking health outcomes to genetic and other molecular measurements at a previously impossible scale, they enable researchers around the world to carry out well-powered experiments. This workshop will offer a hands-on demonstration of some of the issues that may confront first-time users of these enormous resources, including: How to interpret and parse medical outcome data from self-reported assessments, hospital in-patient records, primary care data and other sources. We will demonstrate using an example disease how combining data from different sources substantially increases the power to conduct human disease studies. How to work with 'omics data at this scale. We will use the recently released nuclear magnetic resonance (NMR) metabolomic biomarker dataset to show how additional data types can supplement genotype-phenotype studies. What kind of biases are important to consider in genetic data in hundreds of thousands of samples that are different from smaller studies, and how to use thorough quality control procedures and robust statistical methods to mitigate these. We will use the UK Biobank (https://www.ukbiobank.ac.uk/) to demonstrate these issues, as it has excellent data availability, and can be useful to the widest possible range of new users.

The session will be delivered primarily through interactive Jupyter notebooks, allowing researchers to follow along at their own pace or alongside the instructors. These will be available both in R and Python. We will use mock data and summary statistics similar in structure to UKBB. We will provide all the materials, including simulated datasets, prior to the session, and those will be available online (site, github?). The session will include interactive polling and feedback to better guide the delivery. Reference: https://www.ukbiobank.ac.uk/enable-your-research/approved-research/biomarker-profiling-by-nmr-metabolomics-for-the-study-of-chronic-disease-risk-and-underlying-risk-factors https://www.ukbiobank.ac.uk/enable-your-research/about-our-data/health-related-outcomes-data

Learning Objectives

- Summarize the types of data available in UK Biobank that are relevant to health research.
- Illustrate the strengths and weaknesses of biobank-scale data for genetic epidemiology.
- Contrast analyses that do and do not adjust for confounders.
- Apply best practices in data harmonization to obtain robust, reproducible results.

The Michigan Imputation Server: Data Preparation, Genotype Imputation, and Data Analysis Wednesday, November 1: 2:00 pm – 4:00 pm

Pricing: \$35 ASHG member, \$50 nonmember. **Advance ticket purchase required to attend.** Includes light snacks and drinks.

Speakers: Christian Fuchsberger; Xueling Sim; Saori Sakaue; Sebastian Schoenherr; Lukas Forer; & Albert Smith.

Genotype imputation is a key component of modern genetic association studies. The Michigan Imputation Server has thus far helped > 9,500 researchers from around the world to impute > 95M human genomes. This interactive workshop is intended for anyone interested in learning how to impute genotypes and to use the imputed genotypes, highlighting recent reference panels, including the multi-ancestry panel from the TOPMed program and a specialized HLA panel. A brief overview of imputation and the server will be followed by demonstrations and exercises, including: 1) quality control and preparation of genetic data for use on the MIS with a special focus on diverse ancestries, chromosome X, and the HLA region; 2) tracking runs and use of the application program interface for larger jobs; 3)

downloading data from the MIS and preparing data for genetic analysis; 4) performing a GWAS using imputed data (including HLA region) and interpreting results, taking into account imputation quality; 5) using the additional features, such as the polygenic risk score calculation. We encourage participants to ask specific questions about their own projects. Workshop materials, including slides and example data sets, will be made available before the workshop and will remain online at the MIS website. We expect that this workshop will enable participants to generate high-quality imputed data sets and to effectively analyze them.

Learning Objectives

- Perform quality control and prepare genotype data for imputation.
- Use the imputation server, understand the available parameters, choose the appropriate reference panel, and interpret (error) messages.
- Perform quality control on imputed genotypes, interpret quality metrics, use the imputed genotypes for GWAS, PRS, or meta-imputation.
- Conduct a GWAS analysis using imputed genotypes (with special focus on the HLA region and chromosome X)